ABSTRACT OF THE DISCLOSURE

Mutations resulting in stop codons in the BRCA1 gene are described. All of these mutations result in the formation of a truncated BRCA1 protein. Methods for identifying a sequence variation in a BRCA1 polynucleotide sequence are disclosed. The identification process includes allele specific sequence-based assays of known sequence variations. The methods can be used for efficient, and accurate detection of a mutation in a test BRCA1 gene sample for diagnostic and therapeutic purposes.